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UK Families with Children with Rare Chromosome Disorders: Changing Experiences of Diagnosis and Counseling (2003 to 2013)

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ABSTRACT

The latest United Kingdom (UK) strategy for rare diseases emphasises the need to empower affected populations to improve diagnosis, intervention, and coordination of care. Families who have a child with a rare chromosome disorder (RCD) are a challenging group to include. We report the findings of two large-scale surveys, undertaken by the UK RCD Support Group Unique, of these families' experiences over a ten year period. Seven stages of the patient journey were examined. From pre-testing, through diagnosis, genetics consultation, clinical follow-up and peer support. Overall, 1,158 families replied; 36.4% response rate (2003) and 53.6% (2013). Analysis of responses identifies significant differences ($p < 0.001$) over time with a decrease in results reported face-to-face (76-62%), doubling by telephone (12-22%), improved explanation of chromosome disorder (57-75%), and increased signposting to peer support group (34-62%). However, conduct of the consultation raises a number of important questions. Overall, 28 aspects of the patient journey are recognised as requiring improvement; only 12/28 are currently incorporated in UK service specifications. Involvement of RCD families has identified key service improvements. This approach can empower those affected by such extremely rare disorders, and also enable professionals to design improved services in partnership with 'expert families'. Further surveys are planned.

KEY WORDS

Families' experiences, clinical genetics services, rare chromosome disorders, national surveys, patient reported outcomes (PROs), evidence-based clinical guidelines

1. INTRODUCTION

The United Kingdom (UK) Strategy for Rare Diseases places a strong emphasis on empowering those affected by rare diseases in order to improve diagnosis, intervention, and coordination of care in genetics services¹. Although the strategy was published in late 2013, implementation plans are still being developed for England in 2017 with a view to being fully actioned in 2020. It is acknowledged that this will require “strengthening the mechanisms and opportunities for meaningful and sustained patient involvement in rare disease service provision”.

An important and challenging group to consider when involving those affected by rare diseases will be children with a rare chromosome disorder (RCD). In the UK, it is estimated that at least 300-500 children are born every year with one of a range of RCDs, widely spread geographically². In comparison to more common and well-studied chromosome disorders like Down syndrome, there is far less information available on the natural history or prognosis for these rare diseases (<5 per 10,000 births)³. Their extreme rarity means RCD cases can be particularly challenging for genetics services because, in addition to communicating a laboratory diagnosis, professionals also need to support families who frequently experience severe distress combined with high levels of uncertainty⁴. In such a situation, service providers must ensure that parents understand the diagnosis, help families identify effective coping strategies, and address the lack of available evidence^{5,6}. To date, little is known about the experiences of these families, or the degree to which Clinical Genetics Services currently meet their needs. This is an important gap, since the UK strategy emphasises that successful implementation will require “recognising patient groups as key partners” to develop care pathways that incorporate “best practice from the user perspective”¹.

Patient-reported outcomes for Clinical Genetics Services are still in their infancy⁷. Over the last decade some developments have occurred, largely driven by the extension of clinical genetic services from diagnosing conditions that are exclusively genetic in nature to investigating genetic components for more common diseases⁸, with increased knowledge about the contribution of genetic factors to a range of common diseases⁹. Comprehensive data are not yet available for RCDs although it is anticipated that, in time, the new National Congenital Anomaly and Rare Disease Registration Service established by Public Health England will fill this gap in knowledge¹⁰, with projects such as the Sanger DECIPHER database¹¹ and the Unique registry/database and information guide service¹² also contributing to an improved knowledge base. Currently, there is no European Reference Network (ERN) specific to RCDs despite pan-European efforts to create one. However, the UK-led ERN-Ithaca for intellectual disability and congenital malformations will include RCDs with family support group representation and promises to be a channel through which the experiences of RCD families can be improved.

For chromosome disorders, technological developments such as the introduction of microarray-based comparative genomic hybridisation (*microarray analysis*) techniques have meant that chromosome abnormalities which were formerly too small to be detected by conventional karyotyping can now be identified. Although this has significantly improved sensitivity for detection of clinically relevant genomic imbalances, it has also increased the need for comprehensive genetic counselling to ensure accurate clinical interpretation¹³. In the case of RCDs, clinical interpretation will still face a high level of uncertainty about each affected child's health, potential cognitive development, and life span even after there is a definitive diagnosis¹⁴⁻¹⁶.

In this paper, we present the findings of two large scale surveys which investigated the experiences of UK families who have a child with a RCD over the period 2003 – 2013. We examined the entire care pathway including provision of pre-test information, diagnosis of RCD, genetic counselling, provision of follow-up information and ongoing support. Analysis of responses at different time-points is used to reveal trends and changes over time. The findings should hopefully enable best practice from the user perspective to be more effectively integrated into the implementation phase of the UK Strategy for Rare Diseases.

1.1 Objectives

The study had three main objectives:

1. to examine RCD families' experiences along the entire care pathway;
2. to compare differences over ten years and identify positive or negative changes over time; and
3. to recommend improvements to service provision for this important patient group.

2. METHODS

2.1 Survey Overview

Two surveys were undertaken using a detailed questionnaire designed by Unique, a UK-based Rare Chromosome Disorder Support Group. This support group has over 15,300 member families, representing over 17,000 individuals affected by RCDs, in over 100 countries worldwide, with around 1500 new families registering annually¹⁷. The process for designing the questionnaire is described in Supplementary file 1. The first survey was undertaken in March 2003 and the second in May 2013. Both surveys were limited to members with at least one surviving child with RCD and a valid UK address. The 2013 survey recruited a different cohort of families who joined the group after March 2003. Questionnaires were identical, except for some questions in 2013 relevant to the introduction of microarray analysis¹⁸. The layout of questionnaires was designed to adhere to good practice and minimise the possibility of unreliable data or systematic missing responses¹⁹. Questionnaires in 2003 were pre-printed and posted out to families while questionnaires in 2013 could be completed online or printed off and returned by post; responses were anonymous. The initial invitation in 2013 was followed by two email reminders.

2.2 Questionnaire Content

Questionnaires collected background information on the family. Respondents were then asked about their experiences during different stages of the patient journey (see Table 1). A separate question asked families to rate the quality of the overall service from a user perspective in terms of the overall service received on a ten-point Likert scale ranging from 1 (worst) to 10 (best), and how helpful overall the genetics counselling service has been since their first appointment (4 categories ranging from 'not very helpful' to 'very helpful'). Finally, respondents were invited to give free text descriptions of their experiences of diagnosis and genetic counselling. A copy of the 2003 postal questionnaire is provided in Supplementary file 2.

2.3 Data Analysis

Numerical data were summarised using mean and SD or median and range, depending on data distribution. Analysis was based on completed question responses. There was no imputation of missing data, although we investigated to assess as far as is possible that missing data were missing completely at random. Certain descriptive variables with multiple response categories were dichotomized before analysis e.g. whether person communicating diagnosis was 'genetics professional' or 'non-genetics professional', whether the method of communicating was 'in person face-to-face' or 'indirect by phone, letter etc.'. For comparison of baseline and 2013 responses, chi-square tests were performed for categorical variables and *t*-tests or Mann–Whitney *U* tests for continuous variables. 95% confidence intervals were estimated together with the significance level of observed differences. In addition, some 2013 survey responses were analysed separately for cases diagnosed before and after the introduction of microarray tests in 2008. Stata (version 13) was used for all analyses. Statistical significance was set at $p=0.001$ level.

2.4 Recommended Improvements

A list of recommended improvements was compiled by knowledgeable family members with direct personal experience of RCD, as well as a clinical geneticist and genetics laboratory scientists. Recommendations were based on analysis of data extracted from the questionnaire responses (with detailed examination of levels, significant changes or lack of a significant difference over time).

3. RESULTS

3.1 Respondents

A total of 583/1600 families responded to the 2003 survey (36.4% response rate). In 2013, of 584 responses received; 9 families not resident in the UK were excluded, leaving a total of 575/1072 questionnaires for analysis (53.6 % response rates).

Respondent characteristics were similar in the two groups. Mean age was 42.3 years in 2003 and 43.0 years in 2013. Questionnaires were mainly completed by mothers, although this proportion fell over time from 92.3% to 85.9% in 2013. The majority described themselves as White British/ White European, although ethnic minority respondents doubled over the period from 4.8% to 8.5%. Most families had only

one child with a RCD, with this figure rising over the ten years from 86.1% to 92.3%. A small minority of families had lost a child with a RCD at or after birth; this figure had reduced over time from 6.5% in 2003 to 2.2% in the 2013 sample.

3.2 Rating of service received

When asked how helpful the genetics counselling service had been since their first appointment, Figure 1a shows views were fairly evenly spread across the four categories ranging from 'not very helpful' to 'very helpful', although the most common response was 'had no more contact'. The percentage rating a service as 'not very helpful' did not alter over time; it was 18.7% (95% Confidence Interval (CI): 15.1 - 22.7) in 2003 and 15.0% (95% CI: 11.6 - 19.0) in 2013 ($p=0.161$).

When asked to rate overall service quality on a ten-point scale, average scores rose from 6.37 [SD 2.63] in 2003 to 7.00 [SD 2.52] in 2013. Figure 1b shows mean scores for different professional groups (i.e. genetics doctors, genetics counsellors, and genetics nurses). In 2013, scores were 7.1 (95% CI 6.9, 7.3), 6.5 (6.0, 7.1), and 6.6 (5.7, 7.5) respectively. Therefore, using an unpaired t-test, the genetics doctors scored statistically significantly higher than the genetics counsellors. Comparison of the genetics doctors with the nurses, and of the counsellors with nurses, were not statistically significant.

3.3 Families' experiences over ten years

Families' experiences of services over time are presented in Table 2.

1: Pre-testing process: In 2003 only 70.7% (95% CI: 66.4 - 74.7) of families reported that they had been informed that their child's chromosomes were going to be tested. In 2013, this figure was slightly higher at 73.2% (95% CI: 68.6 - 77.5) but showed no significant improvement over the ten years ($p=0.404$).

2: Test result communication: In 2003, test results were far more likely to be communicated by a paediatrician (64.5% (95% CI: 60.3 - 68.5)) than a genetic specialist (23.8% (95% CI: 20.2 - 27.5)). By 2013, results were almost equally likely to be communicated by a genetic specialist (49.0% (95% CI: 44.3 - 53.7)) or a paediatrician (45.0% (95% CI: 40.3 - 49.7)). Results were rarely reported by other professionals e.g. GPs, genetic nurses, obstetricians, health visitors.

Table 2 indicates that, over the ten year period, it has become significantly less likely ($p<0.001$) that parents will be informed in person about their child's chromosome disorder, although even in 2013 the majority still stated that they were told in person (62.7% (95% CI: 58.2, 67.1)) versus 76.1% (95% CI: 72.4 - 79.6) in 2003. During the same period, communication by telephone doubled from 12.2% (95% CI: 9.6 - 15.2) to 22.1% (95% CI: 18.4 - 26.1) in 2013, and by 50% for letters from 10.1% to 14.4%. Similar trends, away from telling parents in person, are observable for both professional groups (i.e. paediatricians and genetic specialists).

Possibly linked to this trend, responses indicate a shift towards families receiving their test result at home. This has risen significantly ($p<0.001$) from 17.1% (95% CI: 14.0, 20.6) in 2003 to 27.1% (95% CI: 22.9, 31.7) in 2013. At the same time, there has been an increase ($p=0.006$) in test results being communicated in a genetics centre from 8.7% (95% CI: 6.5 - 11.5) to 14.4% (95% CI: 11.1, 18.2); and a significant drop ($p<0.001$) in parents receiving information on the ward after birth or on the children's ward (28.5% (95% CI: 24.6 - 32.5) vs. 14.7% (95% CI: 11.4 - 18.4). Throughout, one in four families continued to receive their test results in a doctor's surgery (26.7% vs. 23.6% in 2013) and one in ten in a child development centre (11.3% vs. 11.5%). The proportion who are told in private has not changed significantly ($p=0.697$) over this period; 80.2% (95% CI: 76.5 - 83.5) in 2003 and 81.1% (95% CI: 77.2 - 84.8) in 2013. Services did not always ensure that support was available from a spouse/partner, relative or friend when imparting this life-changing information. In 2003, one quarter of respondents (23.1% (95% CI: 19.5 - 26.9)) were on their own when they received the diagnosis; rising slightly ($p=0.082$) to 28.0% (95% CI: 23.8 - 32.4)) in 2013. In addition, 47.3% (95% CI: 42.9 - 51.7)) said that their affected child had been present in 2003, and 46.1% (95% CI: 41.3 - 50.9)) in 2013, indicating no significant change ($p=0.710$).

3: Referral to a genetic specialist: Table 2 shows that the proportion of families receiving genetic counselling has decreased slightly ($p=0.031$) from 58.4% (95% CI: 54.2 - 62.5)) in 2003 to 52.0% (95% CI: 47.7 - 56.2)) in 2013. For families informed about their child's test result by a non-geneticist (i.e. paediatrician, GP etc.) likelihood of referral to a genetic specialist has not increased ($p=0.322$), with two out of ten not offered a referral; 22.3% (95% CI: 18.4 - 26.7)) in 2003 and 19.3% (95% CI: 14.9 - 24.2))

in 2013. In families where a second child was diagnosed with an RCD, this figure remains similar (22.7% vs. 21.7% in 2013).

Once referred, the waiting time for an appointment was over 3 months with a slight non-significant ($p=0.105$) increase over time; 95.7 days in 2003 (95% CI: 83.6 - 107.7) and 115.0 days (95% CI: 94.4 - 135.7) in 2013. For a medical geneticist the time has increased from 103.5 days to 172.1 days and for a genetic counsellor from 118 days to 199.1 days. However, for genetic nurses waiting times have fallen from 118 days to 75.6 days. In cases where a definitive diagnosis could not be made at the time of the appointment (15%), further tests are increasingly likely to be ordered; 43.1% of such cases in 2003 and 60.0% in 2013. These further tests produce a change in the provisional diagnosis in one in ten cases (11.6% in 2003 and 9.9% in 2013).

4: Conduct of genetic consultation: Table 2 shows that most respondents considered they had been informed of their child's condition in a sensitive manner; with a slight rise ($p=0.014$) from 66.9% (95% CI: 62.6 - 70.9) in 2003 to 74.1% (95% CI: 69.8 - 78.2) in 2013. There appear to be consistent differences between the perceived sensitivity of different professional groups; for genetic professionals, 81.6% in 2003 and 86.2% in 2013 were viewed as providing the information sensitively, 63.3% and 67.7% of paediatricians respectively, and 43.8% vs. 44.0% of other clinicians.

The conduct of consultations was explored in some detail. Although genetic specialists always introduced themselves (>98% consultations), families were not always told how long the consultation would take (45.6% vs. 38.1% in 2013) or asked what information they already had (24.7% vs. 24.3% in 2013 not asked), and half were not asked how detailed they would like information provided to be (56.5% vs. 54.4% in 2013). Almost half of respondents thought the genetic specialist did not seem to know about them and their family (49.1% vs. 42.3% in 2013), one in five said that a family genetic history was not taken (20.7% vs. 21.0% in 2013), and one third said that there had been no physical examination of their child (37.4% vs. 31.9% in 2013). The risk of having another baby with an RCD was not always explained, with evidence of a decline over time and variation between professionals; medical geneticists (11.8% vs. 20.0% in 2013 did not explain), genetic counsellors (19.0% vs. 32.1%), and genetic nurses (27.8% vs. 30.0%).

Provision of a written summary following the genetic consultation is considered to be good practice²⁰. In 2003, written summaries were provided by 69.0% of medical geneticists, 50.7% of genetics counsellor and 43.8% of genetics nurses. By 2013, although figures had risen to 81.0%, 65.3% and 65.4% respectively, they were still not provided for all as routine practice. On average, families had to wait one month to receive a summary, but some waited as long as 6 months; in 2013 longer delays were reported. The written information provided was considered easy to understand by almost all recipients (93.0% in both 2003 and 2013).

5: Genetic and clinical information provision: Although most respondents could understand the information provided (92.7% in 2003 vs. 89.7% in 2013), the majority considered they had not been given enough information about their child's condition. Table 2 shows this did not change significantly ($p=0.093$) over time; 69.4% (95% CI: 65.2 - 73.4) in 2003 and 64.3% (95% CI: 59.6 - 68.8) in 2013. Responses were not affected by the introduction of microarray analysis, with 66.4% pre-2008 and 61.4% post-microarrays reporting a need for more information. In terms of the content of the information provided, although the majority of families were told which chromosome numbers were involved this has not increased significantly ($p=0.067$); 78.5% (95% CI: 74.6 - 82.0) were told in 2003 and 83.3% (95% CI: 79.4 - 86.7) in 2013. However, explanation of the type of chromosome disorder in a clear and understandable way has improved significantly ($p<0.001$) rising from 57.2% (95% CI: 52.8 - 61.6) in 2003 to 75.1% (95% CI: 70.7 - 79.1) in 2013. Similarly, although a significant proportion of families are not given the karyotype, this has decreased over time (44.4% in 2003 falling to 34.0% in 2013).

Virtually all respondents (95%) said they would have liked a copy of the genetics laboratory report. Although this was not provided in the majority of cases, there is evidence that families are increasingly likely to be given a copy. In 2003, 71.1% were not given a copy, compared to only 48.6% in 2013. However, when laboratory reports were provided, only half included a suitable explanation of the medical or technical terms used, with no evidence of improvement over time (52.2% in 2003 vs. 50.2% in 2013).

In terms of the clinical prognosis, nearly one in three respondents said that they were not told the possible effects on their child of the chromosomal abnormality (30.1% vs. 28.7% in 2013). In cases where this is

provided, accuracy appears to have improved over time e.g. for genetic nurses from 68.3% to 75.7% considered accurate in 2013.

6: Genetic service follow-up: Although most families were offered a further meeting to discuss their child's chromosome disorder, one third reported that they were not. There was a slight but non-significant ($p=0.158$) improvement over time. In 2003, 35.2% (95% CI: 31.1 - 39.5) were not offered a further meeting, falling to 30.9% (95% CI: 26.6 - 35.5) in 2013. Linked to this, only a minority of families said they were told how the genetic counselling service could help them in the future; 28.9% in 2003 and 31.0% in 2013.

7: Signposting to peer support: Signposting of families by all specialists to some form of peer support group has risen significantly, from 34.8% of families in 2003 to 58.7% in 2013. Respondents were increasingly likely to be signposted to Unique (26% in 2003 rising to 67% in 2013). Only a small number of respondents stated that the genetic specialist tried to put them off contacting other affected families (7.3% in 2003 and 3.9% in 2013). Nevertheless, very few respondents (7% in both time periods) reported that they were offered any help to contact other RCD families.

3.4 Recommended improvements identified

Table 3 lists the recommended improvements identified by experts based on survey responses. The penultimate column identifies which are included in the NHS England service specification for organisations funded to provide specialised medical genetics services ²¹. This indicates that 12 out of 28 recommendations identified by the present study are already included in service specifications. However, aspects which are missing include: a) education of non-clinical professionals; b) recommended speed of testing; c) six practical recommendations for communication of test results; d) need to indicate waiting time for referral to a genetics expert; e) five specific recommendations for conduct of consultation with genetics expert. For section f), all recommendations identified by the current study are included in the service specification. The final column shows levels achieved as reported by respondents for selected recommendations included in the service specification. These range from 36% to 80%.

4. DISCUSSION

The necessity for patient reported outcomes (PROs) in clinical genetics services has been identified in a recent review ⁷. However, provision of services to families who have a child with a RCD is acknowledged to be exceptionally challenging ⁴⁻⁶. There is also limited research evidence. A review of research into clinical genetics services and the patient perspective which identified 102 articles ²² found only one focused on these families ³. The recommendations identified in the current study are novel because they are based on the real-life experiences of over one thousand families living with RCDs. As PROs become more important in performance management and funding of health services ⁷, sustained capture of the experiences of such families will be a key challenge ¹. Although a recommendation for 'sustained patient involvement in rare disease service provision' was embedded in the UK strategy for rare diseases, the overall strategy implementation plan for England has only recently been announced ²³.

The large scale surveys reported here show that, although families' rating of service quality has improved over time, key aspects of the 'patient journey' have not and require improvement. Although agreement on key PROs for genetic services is generally acknowledged to be challenging for RCD cases ^{7 24}, our surveys do highlight a number of simple improvements which might be easily introduced and which are indicated elsewhere. For example, a review of guidelines from 18 organisations in six countries on communication of genetic information to families concluded that there was a significant gap in terms of the professional's role in assisting clients to find options for continued support ²⁵. This is a key finding identified from our surveys. Our results also echo evidence from US research which found that parents of children with RCDs were largely disappointed in the counselling they received, although this was only a small-scale study ³.

Although international guidelines for clinical genetics professionals largely cover the professional-client relationship, including respect for the client, maintaining confidentiality, and enabling clients to make informed independent decisions ^{26 27}, they do not include more practical PROs such as those reported in the present study. Other recommendations, such as those produced by Rare Disease UK (a project of the charity Genetic Alliance UK) mostly concentrate on higher level activities (e.g. commissioning and planning of services for rare diseases) with some general recommendations to improve information and support ²⁸. More recent recommendations for reporting the results of diagnostic genetic tests primarily focus on

providing patients with information on how to manage their own condition, something which is less relevant for families of children with RCDs ²⁹. However, the most recent service specification for organisations providing specialised NHS medical genetics services does include some, but not all, of the recommendations identified in the present study ²¹.

Meanwhile, international evidence has emerged of large variations in clinical genetics practice, leading to an increased interest in defining the quality of services and improving delivery models ³⁰⁻³². Core competences and a code of practice have been produced for European health professionals ^{20 33}, based on research by Skirton et al. ³⁴ and approved by the European Society of Human Genetics ³⁵. To date such recommendations are based on the subjective views of professionals, rather than evidence-based, data on user experience. More recently, the US National Society of Genetic Counselors launched a series of new *Evidence-Based Clinical Practice Guidelines* although up to now these do not include RCDs, only Fragile X Syndrome and Down Syndrome. (<http://www.nsgc.org/practiceguidelines>).

Service quality for RCD cases will inevitably be influenced by the availability of genetic specialist expertise. In this respect, the UK appears to be fortunate, with a higher number of genetic counsellors/nurses per million population than other European countries ³². The existence of a long-established Rare Chromosome Disorder Support Group also differentiates the UK from other countries. As the rates of RCD diagnoses rise significantly, thanks to wider use of microarray analyses and the anticipated introduction of next generation DNA sequencing into routine clinical practice, combined with the fact that RCD cases are inevitable geographically widespread, the role of non-geneticist clinicians will inevitably continue at various stages of the patient journey, reinforcing the need for common guidelines, multidisciplinary teamwork, audit checklists, training and coordinated care pathways ^{36 37 38}.

We recognise that the genetic and genomic testing and service landscape in the UK is also developing at a tremendous pace, not least because of the 100,000 Genomes project ³⁹, the creation of 13 Genomic Medicine Centres across the UK ⁴⁰, the Genomics England Clinical Interpretation Partnerships (GeCIP), designed to improve the accuracy and reliability of information fed back to patients ³⁹; and a drive by Health Education England to educate non-genetics healthcare and other professionals in genomic medicine ⁴¹. It is therefore imperative that the value of the expertise of UK families affected by RCDs is not lost in the rapid pace of developments in genomics per se for identifying current and future needs. Although patients' and professionals' views may differ, there does appear to be a level of consensus on important domains such as: decision-making, knowledge of the genetic condition, perceived personal control, risk perception, diagnostic accuracy, and satisfaction/ quality of life ⁴². Also, since clinical genetics services in the UK are currently delivered through a network of 23 centres, this network could facilitate the introduction of a coordinated strategy to support these families, although there is currently no designated centre of excellence specific to RCDs to take the lead. It is possible that an holistic RCD-specific service might be introduced by the newly-emerging rare disease centres, such as those in Birmingham ⁴³ and London ⁴⁴.

Our study inevitably has a number of limitations that should be borne in mind when considering the findings and subsequent recommendations. Firstly, some bias in responses is likely as participants were recruited from a specialist support group and therefore respondents may be different from other UK families with an RCD child. Secondly, it is possible that parents in Unique may be more knowledgeable because they are part of a well-established support group and have higher expectations (e.g. in terms of the information required) than people who do not belong to such an organisation. Thirdly, although the greatest care was taken in questionnaire design, the validity and reliability of the data cannot be tested independently, as with all surveys which record individuals' views¹⁹. Finally, there may be recall inaccuracy since, in some instances, the survey requested information from families sometime after the event.

Conclusions & Recommendations:

These surveys of Unique members address the lack of data on genetic diagnosis and counselling care pathways experienced by families of children with RCDs. Recommendations are offered in the spirit of constructive collaboration to assist clinicians to best meet the needs of patients and their families ²¹. The findings set baseline data for the experiences of families in 2003 and 2013. The intention is to repeat the surveys in 2018/2019 to gather patient-reported experiences as the new streamlined genetics service configuration is rolled out across the UK, and then again in 2021/2022 when new genetics services and implementation plans for the rare disease strategy are well embedded in the UK service provision. We consider that establishment of this form of longer term overview of user experience is particularly important, not least because diagnoses and genetic counselling are likely to be increasingly provided by non-geneticist clinicians.

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CONFLICT OF INTEREST

The authors AS, SW, BS, AK, TP, DB, JE, MH all declare that they have no conflict of interest.

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Figure 1 Rating of Genetic Counselling Services

Figure 1a: Q3.29 How helpful has the genetic counselling service been since your first appointment [%]

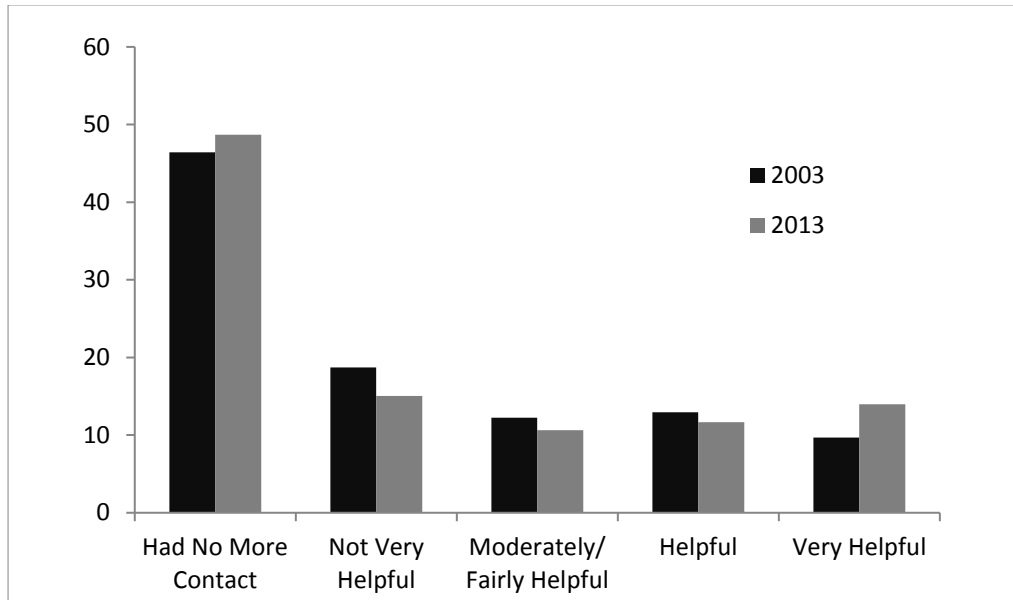
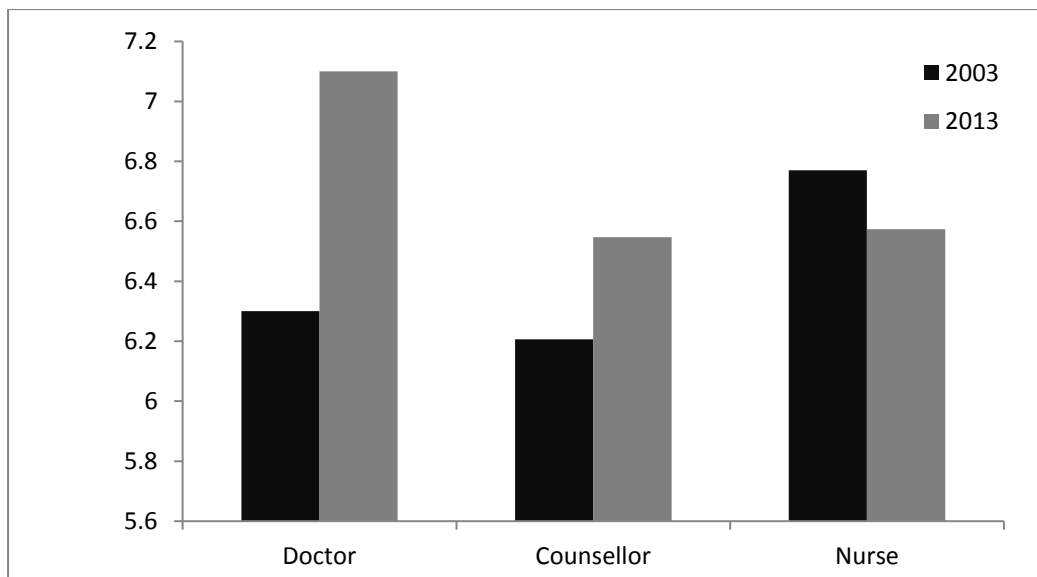


Figure 1b: Q3.32 Overall, how would you rate the service you received from the geneticist(s)** on a scale from 1 (worst) to 10 (best) [Mean Score]



** Genetic specialists include: Medical geneticist, Genetic nurse, Genetic counsellor

Table 1: Questionnaire Content for Stages 1-7 in the Patient Journey

Stages 1-7	Areas Covered in Questionnaire	
Stage 1:Pre-testing process	1.1	Whether family was warned that their child's chromosomes were going to be tested.
Stage 2: Test result communication	2.1	How the diagnosis was communicated e.g. in person, by letter, by telephone etc.
	2.2	Which professional communicated the result e.g. paediatrician, medical geneticist, genetic counsellor, genetic nurse, general practitioner (GP), health visitor, obstetrician, other.
	2.3	Where the respondent was when informed about the test result e.g. in genetics centre, at child development centre, on postnatal ward, on children's ward, at home, at work, in surgery/doctor's room.
	2.4	The time of day, level of privacy and support when result was communicated e.g. whether during the day/evening/weekend, in public/private, on their own or with a spouse or partner/relative or friend present.
Stage 3: Referral to a genetics specialist	3.1	Whether the family was referred to a genetic specialist and, if referred, the waiting time to consultation.
Stage 4: Conduct of genetics consultation	4.1	Conduct of consultation with genetic specialist.
	4.2	Whether a written summary was provided following the consultation.
Stage 5: Genetic and clinical information provision	5.1	Genetics information provided about the chromosome disorder e.g. chromosome number(s) involved, type of disorder, full karyotype.
	5.2	Clinical information provided about the disorder e.g. its rarity, possible effects and details of similar cases.
	5.3	Whether a copy of the cytogenetics lab report was provided and, if not, whether respondent would have liked a copy.
Stage 6: Genetics service follow-up	6.1	Whether a follow-up appointment with a genetics professional was provided.
	6.2	Whether the family was informed about how the genetics service could help them in the future.
Stage 7: Sign-posting to peer support	7.1	Whether the family was given information about support groups.
	7.2	Whether families were encouraged to contact other families similarly affected.

Table 2: Patient Journey - Questionnaire Responses (2003 – 2013)

Question: Information Requested	No. Respondents		2003		2013		Test for difference between percentages (P-value)	Direction of Change*
	2003	2013	Frequency (N)	Percentage (95% CI)	Frequency (N)	Percentage (95% CI)		
	583	575						
1. Warned child’s chromosomes to be tested	474	396	335	70.7 (66.4, 74.7)	290	73.2 (68.6, 77.5)	0.404	NS
2. Results reported by paediatrician	543	447	350	64.5 (60.3, 68.5)	219	49.0 (44.3, 53.7)	<0.001	Decr.
3. Results reported by genetic specialist**	543	447	129	23.8 (20.2, 27.5)	201	45.0 (40.3, 49.7)	<0.001	Incr.
4. How respondent told result - in person	557	467	424	76.1 (72.4, 79.6)	293	62.7 (58.2, 67.1)	<0.001	Decr.
5. How respondent told result - by telephone	557	467	68	12.2 (9.6, 15.2)	103	22.1 (18.4, 26.1)	<0.001	Incr.
6. Place where told - at home	527	416	90	17.1 (14.0, 20.6)	113	27.1 (22.9, 31.7)	<0.001	Incr.
7. Place– genetics centre	527	416	46	8.7 (6.5, 11.5)	60	14.4 (11.1, 18.2)	0.006	Incr.
8. Place– ward after birth/ children’s ward	527	416	150	28.5 (24.6, 32.5)	61	14.7 (11.4, 18.4)	<0.001	Decr.
9. How told result – in private	520	436	417	80.2 (76.5, 83.5)	354	81.1 (77.2, 84.8)	0.697	NS
10. When told result – on your own	520	436	120	23.1 (19.5, 26.9)	122	28.0 (23.8, 32.4)	0.082	NS
11. When told result – affected child/ren present	520	436	246	47.3 (42.9, 51.7)	201	46.1 (41.3, 50.9)	0.710	NS
12. Family received genetic counselling	567	556	331	58.4 (54.2, 62.5)	289	52.0 (47.7, 56.2)	0.031	Decr.
13. Not offered referral to clinical geneticist***	412	296	92	22.3 (18.4, 26.7)	57	19.3 (14.9, 24.2)	0.322	NS
14. Mean time referral to first meeting (days)#	308	279	na	95.7 (83.6, 107.7)	na	115.0 (94.4, 135.7)	0.105	Incr.
15. Family told in sensitive manner	519	437	347	66.9 (62.6, 70.9)	324	74.1 (69.8, 78.2)	0.014	Incr.
16. Family not given enough information when told	517	440	359	69.4 (65.2, 73.4)	283	64.3 (59.6, 68.8)	0.093	NS
17. Explanation of chromosome numbers involved	498	425	391	78.5 (74.6, 82.0)	354	83.3 (79.4, 86.7)	0.067	NS
18. Type of chromosome disorder explained	498	425	285	57.2 (52.8, 61.6)	319	75.1 (70.7, 79.1)	<0.001	Incr.
19. Not offered further meeting after diagnosis	517	434	182	35.2 (31.1, 39.5)	134	30.9 (26.6, 35.5)	0.158	NS
20. Family signposted to peer support group##	475	410	161	33.9 (29.6, 38.3)	254	62.0 (57.0, 66.7)	<0.001	Incr.

* Direction of change. 'Incr.' = Significant Increase (p<0.05); 'Decr.' = Significant Decrease (p<0.05). 'NS' = No significant change (p>0.05).

** Genetic specialists include Medical geneticist, Genetic nurse, Genetic counsellor

*** Families informed about result by a non-genetic specialist (e.g. paediatrician), not offered referral to a clinical geneticist.

Percentage column in this row equals mean time in days.

By medical geneticist

Table 3: Study Recommendations vs NHS England Service Specification, and Levels Achieved.

Stage	Study Recommendation [Link***]	In**	%*
a) Pre-test	<i>Robust procedure, including written, accessible information, to ensure that families are fully informed and truly understand when they consent to their child being referred for genetic testing [1]</i>	Y	73
	<i>Robust procedure, including parents being give information on the possible results from genetic testing including the risk of a non-informative result [1]</i>	Y	73
	<i>Non-genetics clinicians should be educated to recognise when the child's symptoms, including developmental delay, ID, clinical symptoms, behavioural issues etc. indicate that a rare chromosome disorder is a possibility and that genetic testing is appropriate [2, 3]</i>	Y	na
	<i>Non-clinical professionals, e.g. social workers, educators, should be educated to the possibility of a rare chromosome disorder to explain a child's/family's difficulties & to know when to refer families on to a relevant clinician for appropriate assessment for genetic testing [2, 3]</i>	N	-
b) Testing	<i>Genetic testing should be done as quickly as possible, reducing the diagnostic odyssey for these families [14]</i>	N	-
	<i>Parents should be informed how long the results will take to come back & why there might be delays e.g. need for further analysis [1]</i>	N	-
c) Results	<i>Results should be given by a geneticist/competent non-geneticist clinician who fully understands the results & their interpretation & possible implications [2, 3]</i>	Y	na
	<i>Results should ideally be given face to face with the parents [4-11]</i>	Y	63
	<i>Parents' preferences for the means by which the results are given to them should be ascertained at the time of testing [4-11]</i>	N	-
	<i>Results should only ever be given by telephone with the parents' express permission [4-11]</i>	N	-
	<i>Results should never be first revealed in a letter or email, unless requested specifically by parents [4-11]</i>	N	-
	<i>Results should be given in private, either in the clinician's office or at the family home [4-11]</i>	N	-
	<i>Results should ideally be given when both parents are present or if only one parent is available that they be accompanied by a relative or friend for support [4-11]</i>	N	-
	<i>Results should be given to parents without the affected child or other children present [4-11]</i>	N	-
d) Referral	<i>If pathogenic or likely-pathogenic results given by non-geneticist clinician, parents should be offered referral to a clinical geneticist/genetic counsellor [12,13]</i>	Y	80
	<i>Parents should be informed of the waiting time for their appointment with a geneticist [14]</i>	N	-
	<i>Waiting times to see a geneticist should be kept to a minimum to reduce the isolation and anxiety of parents [14]</i>	N	-
e) Consultation	<i>Clinicians should offer the family accessible information about the child's disorder and a prognosis wherever possible e.g. a disorder-specific guide from the Unique website with a clear explanation of any technical/medical terms [16-18]</i>	Y	36
	<i>Clinicians should explain the implications of the diagnosis on parents, siblings and the affected child's future reproductive risks and options, including information on how to explain the diagnosis to the child and siblings where appropriate and how to make sure future genetic counselling is offered [16-18]</i>	Y	36
	<i>All consultations should be conducted in a sensitive, empathetic manner, avoiding jargon and without being rushed [15]</i>	N	-
	<i>Clinicians should introduce themselves to parents and explain how long the consultation will last [15]</i>	N	-
	<i>Parents should be asked what information, if any, they already have about their child's diagnosis and how detailed they want information to be [16 -18]</i>	N	-
	<i>Clinicians should be sure to know at least basic information about the child and the family on first meeting to put the families at ease [15]</i>	N	-
	<i>Clinicians should explain what will happen in consultation, preferably providing families with clear written information beforehand about what to expect [15, 16]</i>	N	-
f) Follow-up	<i>Results should be documented in follow-up letter to parents written in accessible language with a copy of the genetics laboratory report for future reference [16]</i>	Y	36
	<i>Parents should be referred to a reputable support organisation such as Unique for ongoing support, information and networking [20]</i>	Y	59
	<i>Clinicians should offer follow-up meeting if appropriate and/or inform how to contact genetics service with further questions/ need for referral in future [19]</i>	Y	69
	<i>Clinicians should explain what parents can expect from the genetic counselling service in future and any further practical support the service can offer e.g. letters of support explaining the child's disorder to support education/social services/benefits applications [19]</i>	Y	69

Link*** = Question number(s) [1-20] in Table 1 linked to each recommendation.

In** = Whether recommendation currently included (Y/N) in NHS England service specification for organisations funded to provide specialised medical genetics services.

%* = % respondents reporting this was achieved (2013 survey).

Supplementary file 1 – Survey Design

Surveys are designed to explore attitudes, knowledge and behaviours to describe population views and trends. The present surveys come under the category of descriptive surveys where the aim is to explore what is salient to survey participants. Such an approach can provide useful information about social life and services delivery (Bowling, 2009).

In terms of the Unique survey, questions to be included were initially designed by a team of parents drawn from families with RCD(s) and based on matters identified as important to them. Many of these aspects had been highlighted anecdotally by thousands of families registered through Unique's helpline from 1984 to 2003. This is important because, although it is generally not possible to assess validity with a survey, the survey tool will have face validity if respondents recognise questions as pertaining to their experiences.

Following this stage, the actual design of the survey tool was considered very carefully. Questions were designed and set out to try to maximise useful responses. Careful thought was given to mapping the design - using a mix of closed-ended questions and open responses, and using different formats across the survey in order to maximise question completion (Tourangeau, Rips, Rasinski, 2000).

Open response questions were included to provide qualitative data to complement the quantitative data provided by closed-ended questions (reported in the current article). While quantitative analysis of closed question responses can provide focused and highly generalizable information, qualitative data are particularly useful in obtaining an understanding of more subjective and personal aspects (Biggerstaff 2012).

A pilot version of the questionnaire was created which was then trialled by 10 Unique member families plus the core Unique team, along with professional advisors highly experienced in design of questionnaires and of clinical genetics practice.

Following minor revisions, the final questionnaire was disseminated to all relevant UK member families in Unique.

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RARE CHROMOSOME DISORDER SUPPORT GROUP

(Registered Charity No. 1024624)

Survey of *Unique* Families' Experiences of Diagnosis and Genetic Counselling

Dear Unique Member

Our Chief Medical Adviser Professor Maj Hultén has suggested that it would be helpful to find out all our UK members' experiences of diagnosis and genetic counselling via a questionnaire, which is enclosed. The anonymised results will be presented to geneticists at the Annual Conference of the British Society of Human Genetics in York in September, and will also be published in a medical/scientific journal. This is a wonderful opportunity for us to let geneticists know what our experiences have been, good or not so good, and to suggest ways in which the services they provide might be improved.

We would be very grateful if you would please fill in the questionnaire giving as much detail as you can. It should take no more than about 15 minutes of your time. Please try to answer all the questions and return the questionnaire to me in the prepaid envelope provided by 31st March 2003 at the very latest. If you have to leave a question blank please tell us why or write N/A (not applicable). You do not have to give your name and address if you do not want to, although if you do include your details on the sheet provided you will be entered into a prize draw for some travel or Boots vouchers. If you do give your name and address, I will remove these details before the rest of the questionnaire is sent to the University of Warwick for analysis by collaboration with Maj and her colleagues Professor Ala Szczepura and Dr Deborah Biggerstaff.

We value any comments that you are able to make. Please feel free to give any additional information or feedback in the space provided at the end of the survey. All information you give us will remain anonymous and no information will be used which could personally identify you. If you have any questions or would like to talk about the survey, please contact me on 01883 330766. Thank you very much for your help. Your support is very much appreciated and the more of you who reply, the more our opinions and experiences will be listened to.

Best wishes

Beverly Searle

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Families' Experience of Diagnosis/Genetic Counselling

Section 1 – This section is about you and your family

Please answer as many questions as you can and as fully as you can. If you want to clarify any of your answers, please write at the end of the questionnaire.

1.1 Please tell us which adult is completing this form. You don't need to give your name if you do not want to.

(Please choose one category that best describes your family/carer role and tick the relevant box)

- ☐ **Mother**
☐ **Father**
☐ **Female partner**
☐ **Male partner**
☐ **Other** (eg adoptive parent, foster carer).
Please give details

1.2 Please tell us your date of birth

DD MM YY
□□ / □□ / □□

1.3 Please tell us your partner's date of birth

DD MM YY
□□ / □□ / □□

1.4a Please tell us in which part of the country you currently live

County

First part of postcode (eg CR3, RH11, etc)

1.4b Please tell us if you have had any genetic counselling

- ☐ **Yes**
☐ **No**

1.4c If yes, please tell in which part(s) of the country you had any genetic counselling

Town(s) and/or County(ies)

1.5 Is English your preferred language?

(Please tick one box only)

- ☐ **Yes**
☐ **No**

If no, please tell us your preferred language

1.6 Please tell us which group best describes your family?

(Please tick one box only)

- ☐ **African**
☐ **Bangladeshi**
☐ **British/European white**
☐ **Caribbean**
☐ **Chinese**
☐ **Indian**
☐ **Pakistani**
☐ **Other Asian background**
☐ **Other black background**
☐ **Other** (please specify)

Section 2 – This section is about your genetic background

For each question please give your answer by ticking the relevant box or writing on the line

2.1 Are you or your partner a carrier of a chromosomal abnormality?

- ☐ **Yes**
☐ **No**

2.2 If yes, please say which of you is the carrier

2.3a. Do you have any surviving children?

- ☐ **Yes**
☐ **No**

(If no, please go to 2.4)

2.3b If yes, how many surviving children do you have?

□

2.3c How many of your surviving children have been tested for a rare chromosomal abnormality?

□

2.3d How many of your surviving children have a rare chromosomal abnormality?

□

2.3e Please give the date(s) of birth of your surviving child(ren) with a chromosomal abnormality.

Child 1 DD MM YY
□□ / □□ / □□
Child 2 DD MM YY
□□ / □□ / □□
Child 3 DD MM YY
□□ / □□ / □□

2.4 Have you lost any children with a rare chromosomal abnormality at or after birth?

- ☐ **Yes**
☐ **No**

(If no, please go to 2.5)

If yes, please help us by giving some details about the circumstances

Was this child:

- ☐ **Boy**
☐ **Girl**
☐ **Not Known**

Date you lost your child

(please give month and year)

DD MM YY
□□ / □□ / □□

Age of your child (or weeks gestation if baby stillborn) when you lost him/her, if possible

2.5 Female Respondents

Have you had a miscarriage in your present relationship?

- ☐ **Yes**
☐ **No**

Have you had a miscarriage in a previous relationship?

- ☐ **Yes**
☐ **No**

Male respondents

Has your present partner had a miscarriage in your current relationship?

- ☐ **Yes**
☐ **No**

Has a previous partner had a miscarriage?

- ☐ **Yes**
☐ **No**

If you answered yes above:

How many miscarriages have you had?

□

And/or

How many miscarriages has your current partner had with you?

□

How many miscarriages did your previous partner have with you?

□

Section 3 – This section is about your experiences of diagnosis/genetic counselling

3.1 For each person in your family who has a chromosomal abnormality, please give their ages (YY/MM) or the stage of pregnancy (weeks gestation) when diagnosis was first given. For pre-natal diagnosis, please state whether diagnosis was given after testing by CVS and/or amniocentesis. If testing has been done but you have still have no diagnosis, please tell us here. If you lost a baby before birth, please tell us at what stage of pregnancy (weeks gestation) you had reached when you lost your baby:

Mother/female partner

Father/male partner

Child 1

Child 2

Child 3

Baby(ies) lost before birth

3.2 If your child was diagnosed after birth, had you been warned that your child’s chromosomes were going to be tested?

Child 1

1.1

Yes

1.2

No

1.3

Can’t remember

Child 2

2.1

Yes

2.2

No

2.3

Can’t remember

Child 3

3.1

Yes

3.2

No

3.3

Can’t remember

3.3 Who first told you/your partner that your child(ren)/lost baby had a chromosomal abnormality?
(Please tick the relevant professional for each child)

	Child 1	Child 2	Child 3	Lost Baby
Paediatrician (children’s doctor)	<div><div>1.1</div></div>	<div><div>1.2</div></div>	<div><div>1.3</div></div>	<div><div>1.4</div></div>
Clinical geneticist (genetics doctor)	<div><div>2.1</div></div>	<div><div>2.2</div></div>	<div><div>2.3</div></div>	<div><div>2.4</div></div>
Genetics nurse	<div><div>3.1</div></div>	<div><div>3.2</div></div>	<div><div>3.3</div></div>	<div><div>3.4</div></div>
Genetics counsellor	<div><div>4.1</div></div>	<div><div>4.1</div></div>	<div><div>4.3</div></div>	<div><div>4.4</div></div>
GP	<div><div>5.1</div></div>	<div><div>5.2</div></div>	<div><div>5.3</div></div>	<div><div>5.4</div></div>
Health Visitor	<div><div>6.1</div></div>	<div><div>6.2</div></div>	<div><div>6.3</div></div>	<div><div>6.4</div></div>
Obstetrician	<div><div>7.1</div></div>	<div><div>7.2</div></div>	<div><div>7.3</div></div>	<div><div>7.4</div></div>
Other (please specify)	<div><div>8.1</div></div>	<div><div>8.2</div></div>	<div><div>8.3</div></div>	<div><div>8.4</div></div>

3.4 In these following sub-sections, please tell us the circumstances in which you were first informed about your child(ren)’s/lost baby’s chromosomal abnormality(ies).

3.4a How were you told?
(Please tick as many boxes as apply for each of your children/lost baby)

	Child 1	Child 2	Child 3	Lost Baby
On the telephone	<div><div>1.1</div></div>	<div><div>1.2</div></div>	<div><div>1.3</div></div>	<div><div>1.4</div></div>
By letter	<div><div>2.1</div></div>	<div><div>2.2</div></div>	<div><div>2.3</div></div>	<div><div>2.4</div></div>
In person	<div><div>3.1</div></div>	<div><div>3.2</div></div>	<div><div>3.3</div></div>	<div><div>3.4</div></div>
Other (please specify)	<div><div>4.1</div></div>	<div><div>4.1</div></div>	<div><div>4.3</div></div>	<div><div>4.4</div></div>

3.4b Where were you told?
(Please tick one box for each of your children)

	Child 1	Child 2	Child 3	Lost Baby
At home	<div><div>1.1</div></div>	<div><div>1.2</div></div>	<div><div>1.3</div></div>	<div><div>1.4</div></div>
At work	<div><div>2.1</div></div>	<div><div>2.2</div></div>	<div><div>2.3</div></div>	<div><div>2.4</div></div>
In the doctor’s room or surgery	<div><div>3.1</div></div>	<div><div>3.2</div></div>	<div><div>3.3</div></div>	<div><div>3.4</div></div>
On the baby ward/SCBU just after birth	<div><div>4.1</div></div>	<div><div>4.1</div></div>	<div><div>4.3</div></div>	<div><div>4.4</div></div>
On the children’s ward	<div><div>5.1</div></div>	<div><div>5.2</div></div>	<div><div>5.3</div></div>	<div><div>5.4</div></div>
At the Child Development Centre	<div><div>6.1</div></div>	<div><div>6.2</div></div>	<div><div>6.3</div></div>	<div><div>6.4</div></div>
At the genetics centre	<div><div>7.1</div></div>	<div><div>7.2</div></div>	<div><div>7.3</div></div>	<div><div>7.4</div></div>
Other (please specify)	<div><div>8.1</div></div>	<div><div>8.2</div></div>	<div><div>8.3</div></div>	<div><div>8.4</div></div>

3.4c When you were told, was this:
(Please tick as many boxes as apply for each of your affected children/your baby(ies) lost before birth)

	Child 1	Child 2	Child 3	Lost Baby
In private	<div><div>1.1</div></div>	<div><div>1.2</div></div>	<div><div>1.3</div></div>	<div><div>1.4</div></div>
In public	<div><div>2.1</div></div>	<div><div>2.2</div></div>	<div><div>2.3</div></div>	<div><div>2.4</div></div>
During the day	<div><div>3.1</div></div>	<div><div>3.2</div></div>	<div><div>3.3</div></div>	<div><div>3.4</div></div>
During the evening	<div><div>4.1</div></div>	<div><div>4.1</div></div>	<div><div>4.3</div></div>	<div><div>4.4</div></div>
At the weekend	<div><div>5.1</div></div>	<div><div>5.2</div></div>	<div><div>5.3</div></div>	<div><div>5.4</div></div>
When you were on your own	<div><div>6.1</div></div>	<div><div>6.2</div></div>	<div><div>6.3</div></div>	<div><div>6.4</div></div>
With your spouse or partner	<div><div>7.1</div></div>	<div><div>7.2</div></div>	<div><div>7.3</div></div>	<div><div>7.4</div></div>
With another adult relative/friend	<div><div>8.1</div></div>	<div><div>8.2</div></div>	<div><div>8.3</div></div>	<div><div>8.4</div></div>
With your affected child(ren) present	<div><div>9.1</div></div>	<div><div>9.2</div></div>	<div><div>9.3</div></div>	<div><div>9.4</div></div>
With your other child(ren) present	<div><div>10.1</div></div>	<div><div>10.2</div></div>	<div><div>10.3</div></div>	<div><div>10.4</div></div>
Other (please specify)	<div><div>11.1</div></div>	<div><div>11.2</div></div>	<div><div>11.3</div></div>	<div><div>11.4</div></div>

3.5 When you were first told about your child(ren)’s/lost baby’s chromosomal abnormality, what information were you given about the disorder?
(please tick all that apply)

	Child 1	Child 2	Child 3	Lost Baby
Chromosome number(s) involved	<div><div>1.1</div></div>	<div><div>1.2</div></div>	<div><div>1.3</div></div>	<div><div>1.4</div></div>
Chromosome arm	<div><div>2.1</div></div>	<div><div>2.2</div></div>	<div><div>2.3</div></div>	<div><div>2.4</div></div>
Type of disorder (eg deletion)	<div><div>3.1</div></div>	<div><div>3.2</div></div>	<div><div>3.3</div></div>	<div><div>3.4</div></div>
Full karyotype	<div><div>4.1</div></div>	<div><div>4.1</div></div>	<div><div>4.3</div></div>	<div><div>4.4</div></div>
Possible effects of the disorder	<div><div>5.1</div></div>	<div><div>5.1</div></div>	<div><div>5.3</div></div>	<div><div>5.4</div></div>
Rarity	<div><div>6.1</div></div>	<div><div>6.2</div></div>	<div><div>6.3</div></div>	<div><div>6.4</div></div>
Details of other similar cases	<div><div>7.1</div></div>	<div><div>7.2</div></div>	<div><div>7.3</div></div>	<div><div>7.4</div></div>

3.6 When you were first told do you feel you were given enough information?

Child 1

1.1

Yes

1.2

No

Child 2

2.1

Yes

2.2

No

Child 3

3.1

Yes

3.2

No

Baby(ies) lost before birth

4.1

Yes

4.2

No

3.7 When you were first told, do you feel you were told in a sensitive manner?

Child 1

1.1

Yes

1.2

No

Child 2

2.1

Yes

2.2

No

Child 3

3.1

Yes

3.2

No

Baby(ies) lost before birth

4.1

Yes

4.2

No

3.8 When you were first told, were you offered a further meeting at a later date to discuss your child(ren)’s chromosomal abnormality?

Child 1

1.1

Yes

1.2

No

Child 2

2.1

Yes

2.2

No

Child 3

3.1

Yes

3.2

No

Baby(ies) lost before birth

4.1

Yes

4.2

No

3.9 If you were first told about your child(ren)’s chromosomal disorder by a paediatrician, GP or health visitor, did they offer to refer you to a clinical geneticist (ie a genetics doctor), a genetics counsellor and/or a genetics nurse?

Child 1

1.1

Yes

1.2

No

Child 2

2.1

Yes

2.2

No

Child 3

3.1

Yes

3.2

No

Baby(ies) lost before birth

4.1

Yes

4.2

No

3.10 If you were referred to a clinical geneticist (ie a genetics doctor), a genetics counsellor and/or a genetics nurse how long did it take until you actually met that specialist?
(Please specify the number of months or weeks in as much detail as possible in any boxes that apply eg 2M; if less than a month, please write the number of weeks eg 2W)

	Genetics doctor	Genetics counsellor	Genetics nurse
Child 1	<div><div>1.1</div></div>	<div><div>1.2</div></div>	<div><div>1.3</div></div>
Child 2	<div><div>2.1</div></div>	<div><div>2.2</div></div>	<div><div>2.3</div></div>
Child 3	<div><div>3.1</div></div>	<div><div>3.2</div></div>	<div><div>3.3</div></div>
Baby(ies) lost before birth	<div><div>4.1</div></div>	<div><div>4.2</div></div>	<div><div>4.3</div></div>

3.11 Which regional genetics centre or which hospital did the geneticist(s) you saw come from?

3.12 When you first met the clinical geneticist (ie genetics doctor), genetics counsellor and/or genetics nurse did they:
(please tick the yes or no boxes only for each professional you actually saw; leave the other boxes blank. GD - genetics doctor, GC - genetics counsellor, GN - genetics nurse)

Introduce themselves to you?

	GD	GC	GN
Yes	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
No	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
Can't remember	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>

Tell you how long the consultation would take?

	GD	GC	GN
Yes	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
No	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
Can't remember	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>

If other people were present during the consultation, did the geneticist introduce you to them?

	GD	GC	GN
Yes	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
No	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
Can't remember	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>

Ask you what information you had already been given?

	GD	GC	GN
Yes	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
No	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
Can't remember	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>

Seem to know about you and your family?

	GD	GC	GN
Yes	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
No	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
Can't remember	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>

Ask how detailed you wanted any information they gave to be?

	GD	GC	GN
Yes	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
No	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
Can't remember	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>

Give a general explanation of chromosomes/genes?

	GD	GC	GN
Yes	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
No	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
Can't remember	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>

Explain about your or your child(ren)/lost baby's chromosomal abnormality in a clear and understandable way?

	GD	GC	GN
Yes	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
No	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
Can't remember	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>

Give you your own and/or your child(ren)'s karyotype(s)

	GD	GC	GN
Yes	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
No	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
Can't remember	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>

Take a family genetic history (a pedigree)?

	GD	GC	GN
Yes	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
No	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
Can't remember	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>

Make a physical examination of your child(ren)

	GD	GC	GN
Yes	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
No	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
Can't remember	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>

Tell you what the possible effects on your child(ren) of the chromosomal abnormality might be?

	GD	GC	GN
Yes	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
No	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
Can't remember	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>

3.13 How sensitive to your feelings do you think the genetics professional(s) you saw were?
(please tick one option for only the genetics professional(s) you actually saw; leave the rest blank)

Genetics doctor				
Very sensitive	Moderately/ fairly sensitive	Sensitive	A little insensitive	Very insensitive
<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
Genetic counsellor				
Very sensitive	Moderately/ fairly sensitive	Sensitive	A little insensitive	Very insensitive
<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
Genetics nurse				
Very sensitive	Moderately/ fairly sensitive	Sensitive	A little insensitive	Very insensitive
<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>

3.14 How accurate has the information given to you by the genetics professional(s) about the possible effects of your child's chromosomal abnormality turned out to be so far?
(please tick one option for only the genetics professional(s) you actually saw; leave the rest blank)

Genetics doctor				
Very accurate	Moderately/ fairly accurate	Accurate	A little inaccurate	Very inaccurate
<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
Genetic counsellor				
Very accurate	Moderately/ fairly accurate	Accurate	A little inaccurate	Very inaccurate
<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
Genetics nurse				
Very accurate	Moderately/ fairly accurate	Accurate	A little inaccurate	Very inaccurate
<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>

3.15 Did the genetics professional(s) explain to you the risk of you and your partner having another baby with a chromosome abnormality?
(please tick yes or no only for the professionals you actually saw)

Genetics doctor	<input type="checkbox"/>	Yes
	<input type="checkbox"/>	No
Genetics counsellor	<input type="checkbox"/>	Yes
	<input type="checkbox"/>	No
Genetics nurse	<input type="checkbox"/>	Yes
	<input type="checkbox"/>	No

3.16 How well did you understand this explanation?
(please tick one option for only for the genetics professional(s) you actually saw; leave the rest blank)

Genetics doctor				
Very easily	Moderately/ fairly easily	Easily	Not very easily	Not at all
<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
Genetic counsellor				
Very easily	Moderately/ fairly easily	Easily	Not very easily	Not at all
<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
Genetics nurse				
Very easily	Moderately/ fairly easily	Easily	Not very easily	Not at all
<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>

3.17 If either you or your partner carries a balanced chromosomal rearrangement (ie a balanced reciprocal translocation, a Robertsonian translocation, a balanced inversion or a balanced insertion), did the genetics professional(s) offer to help explain to the rest of your family about the possible implications for them?

Genetics doctor	<input type="checkbox"/>	Yes
	<input type="checkbox"/>	No
Genetics counsellor	<input type="checkbox"/>	Yes
	<input type="checkbox"/>	No
Genetics nurse	<input type="checkbox"/>	Yes
	<input type="checkbox"/>	No

3.18a Did the genetic(s) professional(s) tell you about any support groups?

Genetics doctor	<input type="checkbox"/>	Yes
	<input type="checkbox"/>	No
Genetics counsellor	<input type="checkbox"/>	Yes
	<input type="checkbox"/>	No
Genetics nurse	<input type="checkbox"/>	Yes
	<input type="checkbox"/>	No

3.18b Did the genetic(s) professional(s) tell you about Unique?

Genetics doctor	<input type="checkbox"/>	Yes
	<input type="checkbox"/>	No
Genetics counsellor	<input type="checkbox"/>	Yes
	<input type="checkbox"/>	No
Genetics nurse	<input type="checkbox"/>	Yes
	<input type="checkbox"/>	No

3.19 If the genetics professional(s) did not tell you about Unique, how did you find out about us? When you first found out about us, was this before or after you met the geneticist?

3.20a Did the geneticist try to put you off contacting other families affected by a rare chromosomal abnormality?

Genetics doctor	1.1	Yes
	1.2	No
Genetics counsellor	2.1	Yes
	2.2	No
Genetics nurse	3.1	Yes
	3.2	No

3.20b Did the geneticist help you in contacting other families affected by a rare chromosomal abnormality?

Genetics doctor	1.1	Yes
	1.2	No
Genetics counsellor	2.1	Yes
	2.2	No
Genetics nurse	3.1	Yes
	3.2	No

3.20c After meeting him or her, did the genetics professional(s) write to you to explain what you were told at the meeting?

Genetics doctor	1.1	Yes
	1.2	No
Genetics counsellor	2.1	Yes
	2.2	No
Genetics nurse	3.1	Yes
	3.2	No

3.21 If you were sent a letter, how long after your meeting with genetics professional did you receive the letter?

(please write weeks and/or months as accurately as possible in space provided)

Genetics doctor

Genetics counsellor

Genetics nurse

3.23 If the genetics professional(s) did write, was the letter easy to understand?

Genetics doctor	1.1	Yes
	1.2	No
Genetics counsellor	2.1	Yes
	2.2	No
Genetics nurse	3.1	Yes
	3.2	No

3.24 Were you given a copy of your child(ren)'s/your own cytogenetics laboratory report?

- 1 **Yes**
- 2 **No**

3.25 If yes, were you given an acceptable explanation of all the medical or technical words, phrases or codes used in the report?

1 **Yes**

2 **No**

3.26 If no, would you have liked a copy of the report?

- 1 **Yes**
- 2 **No**

3.27 Were you offered a follow-up appointment with the genetics professional(s)?

☐ **1** Yes

☐ **2** No

3.28 Were you told how the genetics counselling service could help you in future?

1 **Yes**

2 **No**

3.29 How helpful has the genetics counselling service been since your first appointment?

Very helpful	Moderately/fairly helpful	Helpful	Not very helpful	No more contact
1	2	3	4	5

3.30 If a diagnosis could not be made at the time of your appointment with the genetics professional:

Have further tests been offered?

1.1 Yes

1.2 No

Have further tests been carried out since then?

2.1 Yes

2.2 No

Have you now been given a diagnosis?

3.1 Yes

3.2 No

3.31 Has the original diagnosis changed?

1 **Yes**

2 **No**

3.32 Overall, how would you rate the service you received from the geneticist(s) on a scale from 1 (worst) to 10 (best)?

(please write in the box(es) your rating for each professional you actually saw)

Genetics doctor	1
-----------------	---

Genetics counsellor 2

Genetics nurse

3.31 Please use the space below to write anything you would like to tell us about your experiences, good or bad, with diagnosis and genetic counselling. From your experiences, have you any constructive suggestions as to how the system/service might be improved? If you need more space please write on extra sheets of paper and attach them securely to this questionnaire.

This image shows a single page of white paper with horizontal ruling lines. The lines are evenly spaced and run across the width of the page. There are no margins, text, or other markings on the paper.[illegible][illegible]This image shows a single sheet of white paper with horizontal ruling lines. The lines are evenly spaced and run across the width of the page. There are no margins, text, or other markings on the paper.

**Now please return this completed questionnaire to
Beverly Searle in the pre-paid envelope provided.**

Please be assured that, as with the rest of this questionnaire, anything you write here will be used entirely anonymously. Thank you for your help and time.

If you would like to enter the prize draw, please enter your name and postal address below. This section will be detached before the rest of the questionnaire is sent on to Professor Maj Hultén and her colleagues.

Your Name

Your Full Postal Address
